



## KIF21A gene

kinesin family member 21A

### Normal Function

The *KIF21A* gene provides instructions for making a protein called kinesin family member 21A. Proteins in the kinesin family are essential for the transport of materials within cells. Kinesin proteins function like freight trains that transport cargo, and their structure is suited for this cargo-carrying function. One end of the protein, called the motor domain, provides power to move the protein and its cargo along a track-like system made from structures called microtubules. The other end of the protein attaches (binds) to specific cargo, such as groups of proteins, for transport. The two ends of each kinesin are connected by a flexible region known as the stalk.

Kinesin family member 21A is found in developing nerve cells (neurons). Researchers believe that this protein carries cargo that is needed for the normal development and function of nerves in the head and face. In particular, this kinesin plays a critical role in the development of cranial nerve III, which emerges from the brain and controls several of the muscles that surround the eyes (extraocular muscles). These muscles direct eye movement and determine the position of the eyes.

### Health Conditions Related to Genetic Changes

#### congenital fibrosis of the extraocular muscles

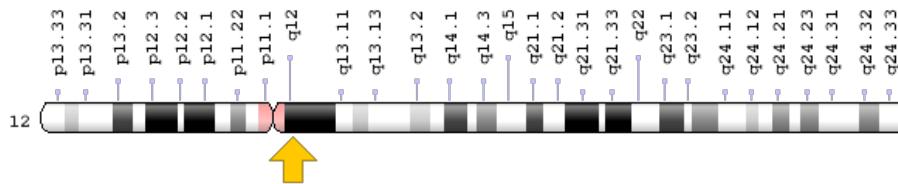
At least 11 mutations in the *KIF21A* gene have been identified in people with congenital fibrosis of the extraocular muscles. These mutations cause the most common form of the disorder, CFEOM1, and are a rare cause of another form of the condition called CFEOM3.

Each of the known *KIF21A* mutations changes a single protein building block (amino acid) in kinesin family member 21A. Most of these changes occur in the stalk region of the protein. These mutations alter the protein's structure, which likely interferes with its ability to transport cargo within neurons. As a result, several cranial nerves and the extraocular muscles they control do not develop normally. Abnormal development and function of these muscles leads to the characteristic features of congenital fibrosis of the extraocular muscles, including restricted eye movement and related problems with vision.

## Chromosomal Location

Cytogenetic Location: 12q12, which is the long (q) arm of chromosome 12 at position 12

Molecular Location: base pairs 39,293,228 to 39,443,414 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- CFEOM
- CFEOM1
- DKFZp779C159
- FEOM
- FEOM1
- Fibrosis of extraocular muscles, congenital, 1, autosomal dominant
- fibrosis of the extraocular muscles, congenital, 1
- FLJ20052
- KI21A\_HUMAN
- KIAA1708
- KIF2
- KIF21A variant protein
- Kinesin-like protein KIF2
- Kinesin-like protein KIF21A
- NY-REN-62 antigen
- Renal carcinoma antigen NY-REN-62

## **Additional Information & Resources**

### Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): There Are Two Types of Microtubule Motor Proteins: Kinesins and Dyneins  
<https://www.ncbi.nlm.nih.gov/books/NBK26888/#A3047>
- Neuroscience (2nd edition, 2001): The Actions and Innervation of Extraocular Muscles  
<https://www.ncbi.nlm.nih.gov/books/NBK10793/>
- The Engle Laboratory, Boston Children's Hospital: KIF21A  
<http://www.childrenshospital.org/research-and-innovation/research/labs/engle-laboratory/neurodevelopmental-research/kif21a>

### GeneReviews

- Congenital Fibrosis of the Extraocular Muscles  
<https://www.ncbi.nlm.nih.gov/books/NBK1348>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28KIF21A%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

### OMIM

- KINESIN FAMILY MEMBER 21A  
<http://omim.org/entry/608283>

### Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=KIF21A%5Bgene%5D>
- HGNC Gene Family: Kinesins  
<http://www.genenames.org/cgi-bin/genefamilies/set/622>
- HGNC Gene Family: WD repeat domain containing  
<http://www.genenames.org/cgi-bin/genefamilies/set/362>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=19349](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=19349)

- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/55605>
- UniProt  
<http://www.uniprot.org/uniprot/Q7Z4S6>

## Sources for This Summary

- Chan WM, Andrews C, Dragan L, Fredrick D, Armstrong L, Lyons C, Geraghty MT, Hunter DG, Yazdani A, Traboulsi EI, Pott JW, Gutowski NJ, Ellard S, Young E, Hanisch F, Koc F, Schnall B, Engle EC. Three novel mutations in KIF21A highlight the importance of the third coiled-coil stalk domain in the etiology of CFEOM1. *BMC Genet.* 2007 May 18;8:26.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/17511870>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1888713/>
- Demer JL, Clark RA, Engle EC. Magnetic resonance imaging evidence for widespread orbital dysinnervation in congenital fibrosis of extraocular muscles due to mutations in KIF21A. *Invest Ophthalmol Vis Sci.* 2005 Feb;46(2):530-9.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/15671279>
- Heidary G, Engle EC, Hunter DG. Congenital fibrosis of the extraocular muscles. *Semin Ophthalmol.* 2008 Jan-Feb;23(1):3-8. doi: 10.1080/08820530701745181. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/18214786>
- Lu S, Zhao C, Zhao K, Li N, Larsson C. Novel and recurrent KIF21A mutations in congenital fibrosis of the extraocular muscles type 1 and 3. *Arch Ophthalmol.* 2008 Mar;126(3):388-94. doi: 10.1001/archophth.126.3.388.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/18332320>
- Marszalek JR, Weiner JA, Farlow SJ, Chun J, Goldstein LS. Novel dendritic kinesin sorting identified by different process targeting of two related kinesins: KIF21A and KIF21B. *J Cell Biol.* 1999 May 3;145(3):469-79.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/10225949>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2185086/>
- Tiab L, d'Allèves Manzi V, Borruat FX, Munier F, Schorderet D. Mutation analysis of KIF21A in congenital fibrosis of the extraocular muscles (CFEOM) patients. *Ophthalmic Genet.* 2004 Dec; 25(4):241-6.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/15621876>

- Yamada K, Andrews C, Chan WM, McKeown CA, Magli A, de Berardinis T, Loewenstein A, Lazar M, O'Keefe M, Letson R, London A, Ruttum M, Matsumoto N, Saito N, Morris L, Del Monte M, Johnson RH, Uyama E, Houtman WA, de Vries B, Carlow TJ, Hart BL, Krawiecki N, Shoffner J, Vogel MC, Katowitz J, Goldstein SM, Levin AV, Sener EC, Ozturk BT, Akarsu AN, Brodsky MC, Hanisch F, Cruse RP, Zubcov AA, Robb RM, Roggenkämper P, Gottlob I, Kowal L, Battu R, Traboulsi EI, Franceschini P, Newlin A, Demer JL, Engle EC. Heterozygous mutations of the kinesin KIF21A in congenital fibrosis of the extraocular muscles type 1 (CFEOM1). *Nat Genet*. 2003 Dec; 35(4):318-21. Epub 2003 Nov 2.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/14595441>
- Yamada K, Chan WM, Andrews C, Bosley TM, Sener EC, Zwaan JT, Mullaney PB, Oztürk BT, Akarsu AN, Sabol LJ, Demer JL, Sullivan TJ, Gottlob I, Roggenkämper P, Mackey DA, De Uzcategui CE, Uzcategui N, Ben-Zeev B, Traboulsi EI, Magli A, de Berardinis T, Gagliardi V, Awasthi-Patney S, Vogel MC, Rizzo JF 3rd, Engle EC. Identification of KIF21A mutations as a rare cause of congenital fibrosis of the extraocular muscles type 3 (CFEOM3). *Invest Ophthalmol Vis Sci*. 2004 Jul;45(7):2218-23.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/15223798>

---

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/gene/KIF21A>

Reviewed: March 2009

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications

U.S. National Library of Medicine

National Institutes of Health

Department of Health & Human Services